

## Supplemental Data

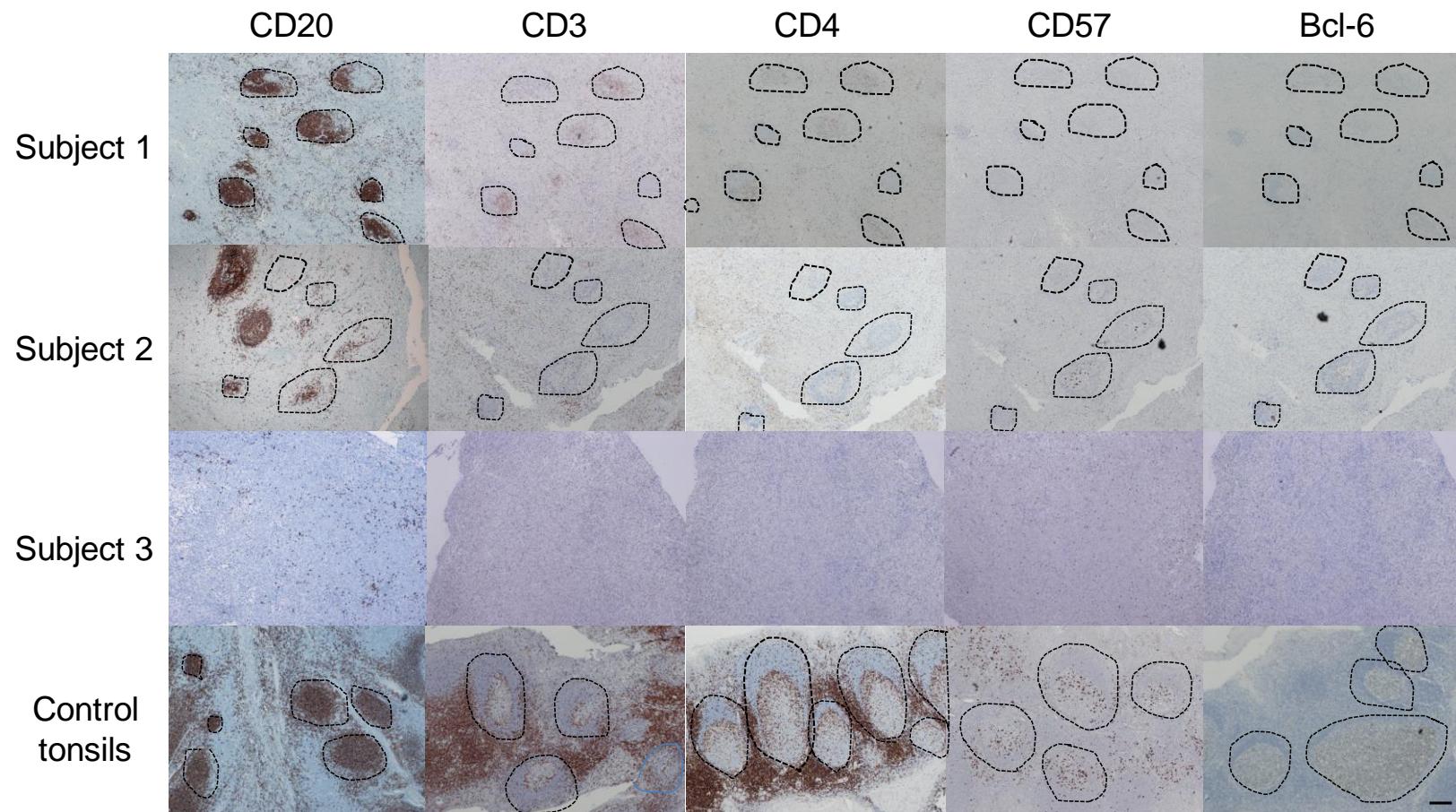
### IL-12 Receptor $\beta 1$ Deficiency Alters *in vivo* T Follicular Helper Cell Response in Humans

Nathalie Schmitt<sup>1</sup>, Jacinta Bustamante<sup>2</sup>, Laure Bourdery<sup>1</sup>, Salah Eddine Bentebibel<sup>1</sup>, Stephanie Boisson-Dupuis<sup>3</sup>, Fran Hamlin<sup>4</sup>, Mau V. Tran<sup>4</sup>, Derek Blankenship<sup>1</sup>, Virginia Pascual<sup>1</sup>, Daniel A. Savino<sup>4</sup>, Jacques Banchereau<sup>1</sup>, Jean-Laurent Casanova<sup>2,3</sup>, and Hideki Ueno<sup>1</sup>

#### Supplementary Figure 1-4

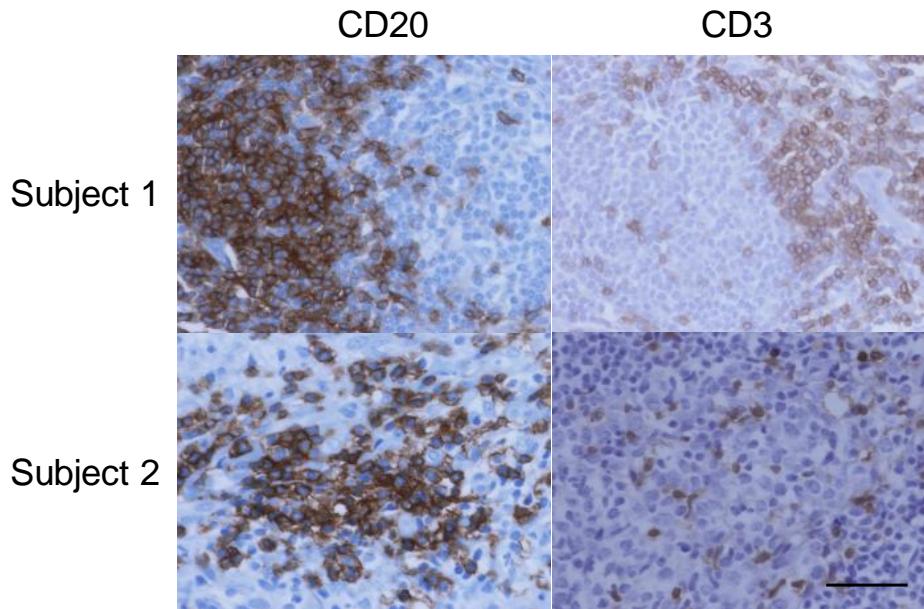
#### Supplementary Table 1

**Fig. S1: Low magnification images of immunohistochemistry staining of LNs from IL-12R $\beta$ 1-deficient subjects**



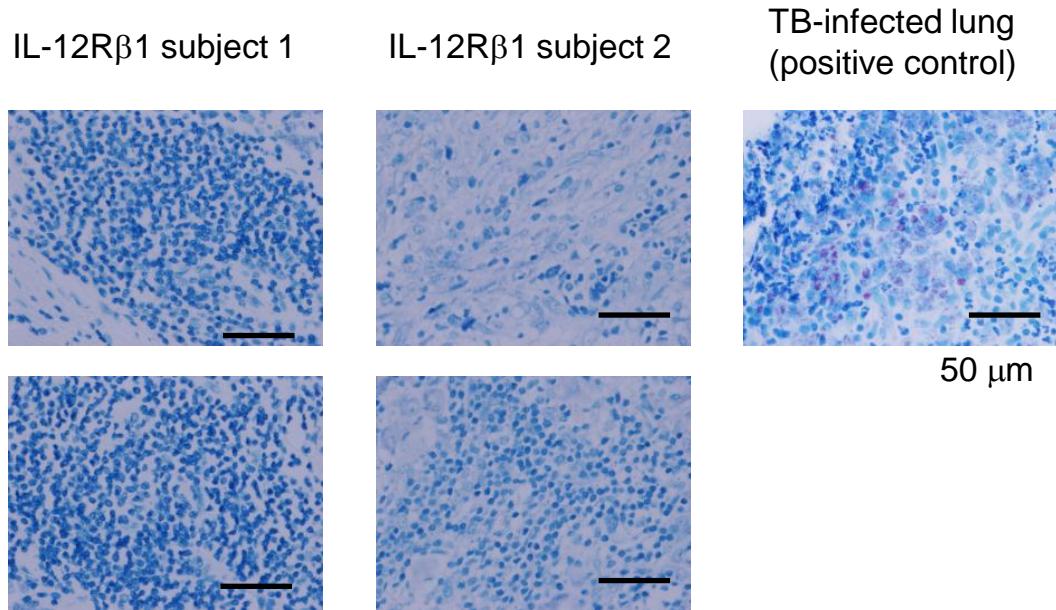
Immunohistochemistry staining of peripheral LN samples from IL-12R $\beta$ 1-deficient subjects and tonsil samples from control subjects. Samples were stained CD20 mAb, CD3 polyclonal Ab, CD4 mAb, CD57 mAb and Bcl-6 mAb. T-B cell aggregates in subject 1 and 2, and secondary follicles in control tonsils were indicated by dotted lines. Bar equals 200  $\mu$ m.

**Fig. S2: High magnification images of T-B cell aggregates in LNs from IL-12R $\beta$ 1-deficient subjects**



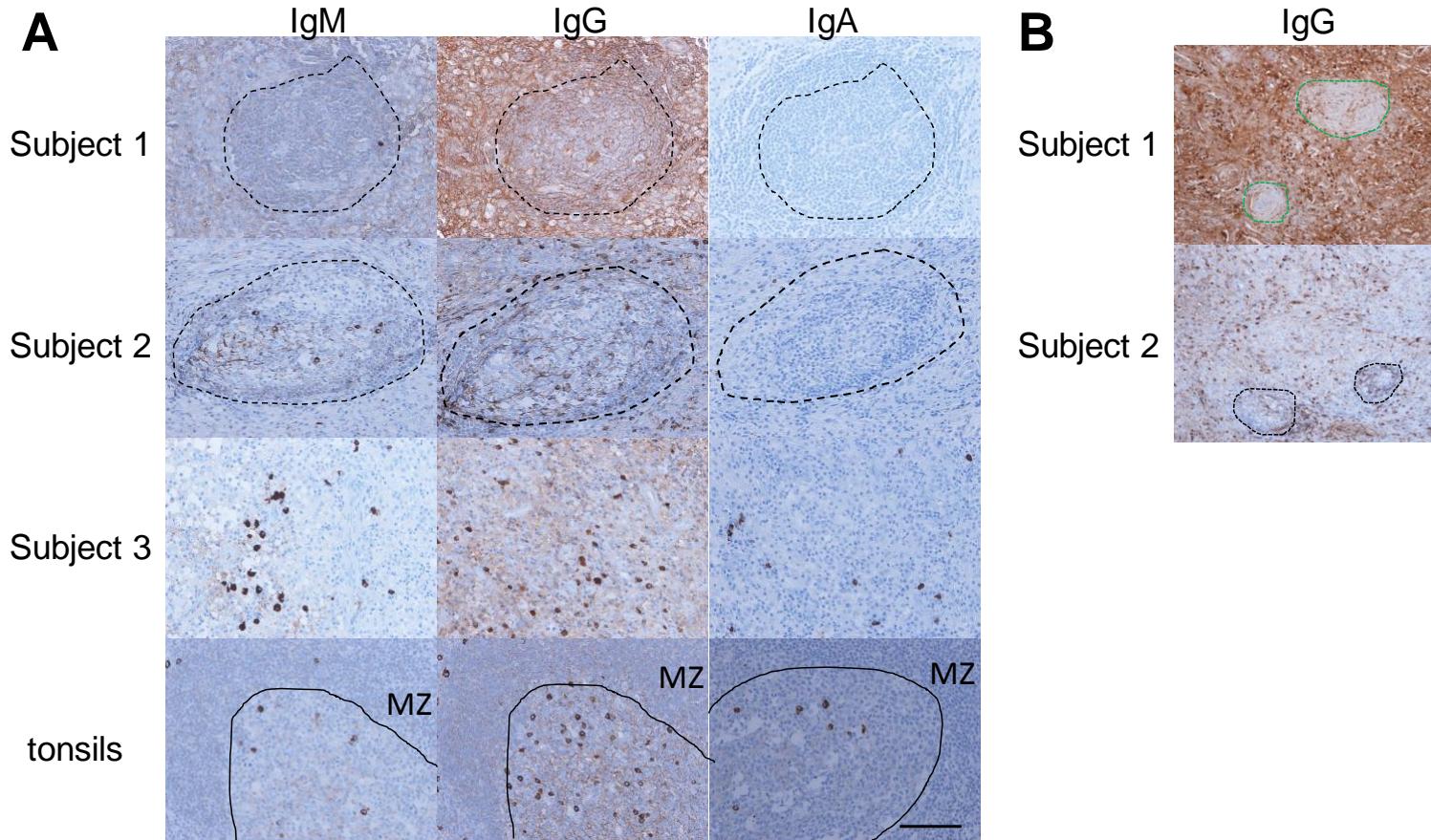
Immunohistochemistry staining of peripheral LN samples from IL-12R $\beta$ 1-deficient subjects. Samples were stained CD20 mAb and CD3 polyclonal Ab. High magnification images of T-B cell aggregates (indicated by dotted lines in Figure 4) are shown. Bar equals 100  $\mu$ m.

**Fig. S3: T-B cell aggregates in LNs from IL-12R $\beta$ 1-deficient subjects did not contain BCG.**



The presence or absence of acid-fast bacillus in the T-B aggregates in LNs of IL-12R $\beta$ 1-deficient subjects (indicated by dotted lines in Figure 4) was determined by Ziehl-Neelsen staining. The positive control staining is shown at the right panel.

**Fig. S4: Ig staining of LNs from IL-12R $\beta$ 1-deficient subjects**



Immunohistochemistry staining of peripheral LN samples from IL-12R $\beta$ 1-deficient subjects. Samples were stained IgM, IgG, and IgA mAbs. **A.** T-B cell aggregates in subject 1 and 2 are indicated by dotted lines. In control tonsils, the margin between the mantle zone

(MZ) and the GC are indicated by solid lines. Bar equals 200  $\mu\text{m}$ . **B.** Lower magnification images of IgG staining in LN samples from IL-12R $\beta$ 1-deficient subjects. Green dotted lines in subject 1 indicate granulomas. Black dotted lines in subject 2 indicate the border of T-B cell aggregates.

**Table S1: The list of control and IL-12R $\beta$ 1-deficiency subjects**

Table S1A: PBMC donors. *IL12RB1* mutation is indicated.

	Sample ID	gender	age (years)	Mutation
IL12R $\beta$ 1 deficiency	IL12RB1-1	F	7 and 12	783+1G>A
	IL12RB1-3	M	4 and 5	700+362_1619-944del
	IL12RB1-4	F	26	[1745_1746delinsCA]+[1483+182_1619-1073del]
	IL12RB1-5	F	4	1623_1624delinsTT
	IL12RB1-6	F	6	549+2T>C
	IL12RB1-7	F	2	[1440_1447delins16]+[Q171P]
	IL12RB1-8	F	24	[C196Y]+[1483+182_1619-1073del]
	IL12RB1-9	F	1	711insC
	IL12RB1-10	M	3	E480X
	IL12RB1-11	F	13	1791+2T>G
	IL12RB1-12	M	16	R213W
	IL12RB1-13	M	15	C198R
	IL12RB1-15	F	0.5	700+362_1619-944del
	IL12RB1-16	F	3	Q32X
	IL12RB1-17	F	12	781+1G>A
	IL12RB1-18	F	7	781+1G>A
	IL12RB1-19	M	1	1791+1G>A
	IL12RB1-2	M	35	1623_1624delinsTT
	IL12RB1-20	F	6	C198R
	IL12RB1-21	F	2	1791+2T>G
	IL12RB1-22	F	6	1791+2T>G
	IL12RB1-23	M	2	[E67X]+[1623_1624delinsTT]
	IL12RB1-24	F	19	1791+2T>G
	IL12RB1-25	F	5	1791+2T>G
Control	H370	M	2	
	H322	M	2	
	H369	M	3	
	H385	M	3	
	H361	F	3.5	
	H320	F	4	
	H239	M	5	
	H251	F	5	
	H322	M	5	
	H374	F	6	
	H367	M	7	
	H378	M	7	
	H240	M	8	
	H274	M	9	
	H288	F	9	
	H288	F	9	
	H366	M	9	
	H368	M	9	
	H220	M	9	
	H252	M	9	
	H241	F	10	
	H248	M	10	
	H375	M	10	
	H377	F	10	
	H382	M	10	
	H384	F	11	
	H287	M	11	
	H242	M	12	
	H250	M	12	
	H383	M	12	
	H319	M	12	
	H287	M	13	
	H381	M	13	
	H217	F	14	
	H290	F	14	
	H271	M	15	
	H380	F	15	
	H170	M	16	
	H302	M	16	
	H364	F	16	
	H243	F	17	
	H359	F	17	
	3145058	F	20	
	3144223	F	21	
	3144274	F	21	
	6405003	M	22	
	314459	F	22	
	3144581	M	23	
	3143861	F	24	
	3143896	F	25	
	3144424	M	25	
	C6	M	26	
	3144266	M	27	
	C1	F	27	
	5541898	F	28	
	C7	M	28	
	3144442	F	29	
	C2	F	29	
	C3	M	29	
	C4	M	30	
	C8	F	30	
	3145254	F	33	
	C5	F	33	
	314467	M	35	
	3144813	M	35	

Table S1B: Serum donors

	<b>Sample ID</b>	<b>gender</b>	<b>age (years)</b>	<b>Mutation</b>
IL12R $\beta$ 1 deficiency	S-1b	F	6	C198R
	S-2b	F	11	1791+2T>G
	S-4b	F	14	Q32X
	S-5b	F	16	Q32X
	S-6b	M	2	700+362_1619-944del
	S-7b	F	26	[1745_1746delinsCA]+[1483+182_1619-1073del]
	S-8b	F	14	781+1G>A
	S-9b	F	19	781+1G>A
	S-10	M	29	1623_1624delinsTT
	S-11b	F	1	711insC
	S-12b	M	11	1021+1G>C
	S-14b	M	11	R173P
	S-15b	F	24	[C196Y]+[1483+182_1619-1073del]
	S-16b	F	2	[I369T]+[1623_1624delinsTT]
	S-19	M	8 and 11	Y88X
	S-20	M	4	Y88X
	S-21	M	2	G478R
	S-22b	M	1	64+1G>T/64+1G>T
	S-24b	M	26	K305X
	S-25b	F	2	1791+2T>G
	S-26b	F	11	700+362_1619-944del
	S-27b	M	33	1765delG
	S-28	F	15	1623_1624delinsTT
	S-29	F	12	1623_1624delinsTT
	S-33	M	15	700+362_1619-944del
	S-34	M	14	700+362_1619-944del
	S-35	F	2	R213W/R213W
Control	S-3143896	F	25	
	S-314459	F	22	
	S-314467	M	35	
	S-C3	M	29	
	S-C5	F	33	
	S-H218	F	11	
	S-H220	M	8	
	S-H225	F	9	
	S-H228	F	7	
	S-H230	F	14	
	S-H252	M	8	
	S-H265	F	5	
	S-H287	M	11	
	S-H288	F	7	
	S-H290	F	12	
	S-H295	M	5	
	S-H298	F	10	
	S-H301	F	16	
	S-H307	F	12	
	S-H311	F	17	
	S-H315	M	11	
	S-H317	M	16	
	S-H318	F	15	
	S-H322	M	2	
	S-H325	M	13	
	S-H341	F	6	
	S-H342	M	6	
	S-H345	F	5	
	S-H361	F	3	
	S-H50	F	14	
	S-H59	F	19	